

Illinois Department of Public Health
Genetic and Metabolic Disease Advisory Committee (GMDAC)
Minutes – October 27, 2011
Hilton Garden Inn, Springfield, IL

Members Present:

Joel Charrow, M.D., Chair, Children's Memorial Hospital
Darrel Waggoner, M.D., University of Chicago
Susanna McColley, M.D., Children's Memorial Hospital
Michael Schneider, M.D., SIU School of Medicine
Lainie Friedman Ross, M.D., Ph.D., University of Chicago Hospital
Cathy Gray, R.N., University of Chicago
Margaret Westemeyer, M.S., University of Illinois at Peoria
Colleen Gibson, R.N., LaSalle County Health Department
Sheila Chalmers-Currin, Parent
Sean Clark, Parent
Barb Goss, R.D., University of Illinois at Chicago

GMDAC Members Not Present:

Karen Litwack, M.S.W.	W. Patrick Zeller, M.D.
George Hoganson, M.D.	Praveen Kumar, M.D.
John Ralston, M.D.	Cathy Wicklund, M.S.
Alexis Thompson, M.D.	Albert Olorvida, M.D.
Michael Msall, M.D.	

Other Attendees:

Annemarie Valdez, SIDS of Illinois
Talana Hughes, Sick Cell Diseases Association of Illinois (SCDAI)
Amy Walsh, Parent
Zina Berryhill, Parent
Tess Rhodes, R.N., Division of Specialized Care for Children (DSCC)
Susan Gentili, Graduate student intern with DSCC

IDPH Representatives:

David Culp, Ph.D., IDPH Deputy Director
Tom Schafer, IDPH Deputy Director
Tom Johnson, Division Chief IDPH Laboratories
Matt Charles, Asst. Division Chief IDPH Laboratories
George Dizikes, Ph.D., Chief IDPH Newborn Screening Laboratory
Mike Petros, Dr. P.H., Operations Manager, IDPH Newborn Screening Laboratory
Jane Fornoff, Ph. D., IDPH Adverse Pregnancy Outcomes Reporting Service (APORS)
Elizabeth Patton, J.D., IDPH Legal Services
Gail Tanner, Au. D.

Genetics/Newborn Screening Program:

Claudia Nash, Program Administrator	Debbie Box	Elaine Huddleston
Nikki Woolverton, Grants Manager	Denise Kink	Margie Nelson
Barbara DeLuka, Nurse Consultant	Laura Harris	Angela George
Heather Shyrock, Operations Manager	Karen Devine	Dennis Tiburzi
Doug Hart	Lori Bless	Rhonda Hawkins

Introductions and Comments:

The meeting was called to order at 10:20 a.m. Following introductions, the minutes of the March 31, 2011 Spring meeting were approved.

Bylaws – Review of Changes:

Claudia Nash discussed appointments and term limits for members, and proposed changes to the GMDAC bylaws, including the addition of a provision for members to attend meetings by teleconferencing or video conferencing. Addition of the Newborn Screening Expansion Subcommittee as a “standing” subcommittee, rather than an ad hoc subcommittee was also suggested, if approved by the GMDAC members. A motion was made by Dr. McColley to approve the new provisions to the bylaws; this was seconded by Barb Goss, and approved by the committee members.

Subcommittee Reports:

Newborn Screening and Laboratory Subcommittee (NSLS)

In the absence of the Subcommittee Chair, Dr. Hoganson, Dr. Petros discussed the minutes of the September 28, 2011 Newborn Screening and Laboratory Subcommittee meeting; a copy of the minutes was provided to the members. Laboratory staffing, and utilization of and improvements to the newborn screening data system were discussed. A pilot project between the IDPH newborn screening laboratory and Northwestern Memorial Hospital for electronic HL7 data transfers between the two facilities was announced, and following successful completion of this pilot, options for electronic data transfers of demographic data and laboratory test results will be offered to other perinatal hospitals. Recent changes to the galactosemia screening and proposed changes to the DNA mutation panel kits for cystic fibrosis screening were also discussed. The Clinical Laboratory Standards Institute (CLSI) guidelines (published in 2009) for newborn screening in the NICU population and data collected on confirmed hypothyroidism cases were reviewed by the NSLS and a recommendation was made by the Subcommittee to follow CLSI guidelines and add a third routine newborn screening sample collection for newborns with birth weights less than 2,000 grams at day 28 of life or prior to discharge from the NICU, whichever is first. It was acknowledged that implementation of the new requirement will require changes to the Newborn Screening Administrative Rules.

Newborn Screening Expansion Subcommittee (NSES)

Dr. Waggoner discussed the October 3, 2011 meeting of the NSES and the minutes were provided to the GMDAC members. Kathleen Sebelius, the Secretary of the Department of Health and Human Services, and the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children’s (SACHDNC) approved the addition of pulse oximetry for critical

congenital heart disease (CCHD) as the thirtieth disorder to be added to the uniform panel of newborn screening disorders for all states. Following review and discussion among the NSES members, the Subcommittee voted that a recommendation be made to the Director of Public Health by the GMDAC to add CCHD to the Illinois newborn screening panel. While the voting margin for the recommendation was narrow, it was acknowledged the cost to hospitals and the logistics of providing the screening and follow-up were the major concerns among members not voting in favor of the motion. It was felt that the SACHDNC review had been very thorough and lengthy and that the evidence for the effectiveness of the screening in detecting children with congenital heart defects was convincing. Dr. Waggoner stated that the NSES members felt a recommendation for the addition of CCHD, also necessitated a recommendation for establishing a working group of pediatric cardiologists, neonatologists, hospital representatives and other interested parties to assist in planning for possible implementation of this hospital based point of service screening. It was noted that a GMDAC member and a NSES member were both on the SACHDNC work group that evaluated the nomination of CCHD for inclusion in the uniform screening recommendations and created guidelines for implementation. Division of Specialized Care for Children will provide third party payment for diagnostic testing and some treatment services for newborns identified with CCHD, as heart defects are included in the recognized criteria for services provided by this agency. DSCC has an established network of providers and services for children with heart diseases.

A motion for recommendation of CCHD screening and creation of a working group to assist in implementation to the Director of Public Health was made by Cathy Gray, seconded by Dr. McColley and approved by the members of the parent Committee, with one objection by Dr. Schneider noted. Dr. Ross abstained.

Cystic Fibrosis (CF) Collaborative:

Dr. McColley provided a power-point presentation on the work of the CF Collaborative and information about the Cystic Fibrosis Foundation (CFF) quality improvement grant. All CF Centers are participants in the Collaborative, which was established to assure timely, efficient, and effective diagnostic testing for newborns with positive screens for CF, and to provide genetic counseling services for parents. The treatment goal for CF is to improve early childhood nutrition, as improved early nutrition and growth are good predictors for better pulmonary outcomes in CF patients. The Collaborative is working to reduce the number of "quantity not sufficient" (QNS) diagnostic sweat tests performed at each Center, and the Director of Children's Memorial laboratory is available to any CF Centers that would like assistance in reducing QNS rates. The recently approved CFF grant funding has also enabled Centers to request site visits from Dr. LeGrys of the CFF.

Newborn Screening Fee Utilization:

Claudia Nash provided information on the fees charged by other states for newborn screening. Illinois provides screening and a greater level of services than many of the states charging even higher fees for newborn screening. The fee will be increased from \$78 to \$88 to provide equipment and staff for implementation of screening and follow-up services for severe combined immune deficiency (SCID). There was discussion about funding issues for

laboratory equipment, laboratory and follow-up program staffing, and additional infrastructure needs to collect data and provide follow-up services for critical congenital heart disease, should this condition be added. Members voiced concerns about future metabolic fund sweeps, but were reassured additional sweeps of this dedicated fund are unlikely at this time.

Lysosomal Storage Disease (LSD) Implementation Update:

Dr. Dizikes provided a detailed handout on the validation plan for tandem mass multiplex LSD assays by spectrometry screening (MS/MS) which could begin in April of 2012. CLIA approval of the PerkinElmer, Inc. reference lab testing for six of the seven LSD disorders legislated for Illinois newborn screening will provide a means for validating and reporting results through a contractual agreement whereby samples could be overnight shipped to Perkin-Elmer for testing and reporting, while IDPH lab tests the samples internally and verifies results with PerkinElmer, Inc. Entry of demographic information, recording of results and case creation would be accomplished using HL7 data transfers between the PerkinElmer, Inc. and IDPH Lifecycle and Patient Care data systems currently in use at the laboratory and the follow-up program. The LSD assays currently available through PerkinElmer, Inc. include: Pompe, Fabry, Gaucher, Krabbe, Niemann-Pick, and MPS1 (Hurler's), with MPS2 (Hunter's) expected to become available in January 2012. Additional MS/MS instruments will be required to initiate LSD screening due to the age of existing instruments and the need for high through-put of samples for statewide LSD screening.

It is hoped that IDPH can conduct two pilots, one during the spring and another immediately following, during the summer. This dual piloting would allow for evaluation of temperature sensitivity of the LSD enzymes; the initial use of de-identified samples for staff training; and parallel testing with PerkinElmer, Inc. and the IDPH Division of Laboratories. It is anticipated that Northwestern Memorial Hospital and the University of Chicago Hospital would participate in these phase-in pilots utilizing a memorandum of understanding (MOU) between IDPH and the hospitals. The MOU would address confidentiality concerns of all parties and residual newborn screening samples would be returned to IDPH by PerkinElmer, Inc.

There was discussion among the members concerning the need for informed consent of parents to provide screening for LSD's. Some published studies have indicated difficulties in determining age at disease onset for Pompe and Fabry, and concerns were voiced about the disease risk and the uncertain effects of Krabbe, as well as treatment options and outcomes for this disorder. Dr. Ross stated she feels that while screening has been legislatively mandated, the federal regulations for the protection of human subjects requires IRB-approval and informed consent for research. Given the many unknowns for screening for lysosomal storage diseases, it fails to meet the Wilson and Jungner criteria and therefore is better classified as research. Federal law would require that screening be classified as research, and thus would require an IRB-protocol with parental consent and greater flexibility to refuse to enroll a child in research. Dr. Ross suggested that review by the IDPH Institutional Review Board should be considered prior to

start up of the LSD phase-in pilot, although LSD screening has been legislatively mandated for statewide screening. This will be taken into consideration by IDPH representatives.

Severe Combined Immune Deficiency Implementation Update:

Dr. Dizikes also provided a detailed handout for validation of SCID screening utilizing a MOU with either Wisconsin State Laboratory of Hygiene (WSLH) or PerkinElmer, Inc. Both facilities are CLIA approved for SCID testing. In either agreement, IDPH would send samples to the responsible laboratory for testing and reporting of results, while IDPH lab performs parallel testing for validation. The testing for SCID is real time PCR, and involves quantification of small DNA fragments called T-Cell Excision Circles (TRECs), which are a by-product of normal immune system development in newborns. This screening has been implemented in several states and has been shown to be effective with a low false positive rate. Equipment and staff training will be necessary, as well as contractual arrangements with WSLH or PerkinElmer, Inc., prior to start-up of the phase-in pilot, to possibly occur within about 6 months.

Newborn Screening Follow-Up Program

Data on confirmed cases and abnormal screen cases for calendar year 2011 (included in packet) was provided; to date 229 infants have been diagnosed with a newborn screening disorder this year, and additional cases are still pending definitive diagnosis. Over 140,000 specimens have been received and tested during this period, and over 10,000 newborns have been followed for abnormal newborn screening results. Follow-up program staff are working more closely with other IDPH Divisions including APORS Birth Defects Registry and Vital Records, and with other agencies including Department of Children and Family Services in an effort to improve both short-term and long-term follow-up services for children.

A quality assurance report for perinatal hospitals was provided. During the third quarter of 2011, 77.9% of specimens were received by the Chicago laboratory within 3 days of collection, and 98.8% were received within 5 days, with 99.9% of specimens received as satisfactory for testing. The assistance of the IDPH Perinatal Network Administrators in promoting newborn screening and utilizing newborn screening quality assurance data in perinatal hospital evaluations was acknowledged.

A survey of other states with regards to CCHD screening was provided and it was acknowledged that most states are struggling to develop reporting and follow-up services for CCHD. These services will require coordination and cooperation between hospitals and IDPH, quite possibly relying on the assistance of other IDPH Divisions and programs, including the APORS, Perinatal Network and Newborn Hearing Screening program. Information about the incidence of diagnosed cases of congenital hypothyroidism (CH) among low birth weight newborns was provided. In 2006 IDPH instituted a routine second newborn screen for newborns admitted to a NICU. From 2006 through 2010, the number of cases of CH with a normal initial screen at 24 hours of life and a second normal screen at 14 days has been higher than expected. Information about Clinical Laboratories Standards Institute national guidelines for newborn screening in the NICU has been presented at previous GMDAC

meetings, and as mentioned in the NSLS subcommittee report, the NSLS has made a recommendation to follow the CLSI guidelines and require a routine third newborn screen to be collected from all newborns with birth weights less than 2000 grams at day 28 of life, or prior to discharge from the NICU. There were no objections to the NSLS recommendation from GMDAC members.

Genetic Grantee Update:

Nikki Woolverton reported on grants provided to medical centers and local health departments providing genetic services and to pediatric hematology services for individuals with sickle cell diseases. For fiscal year 2012, IDPH is providing funding for sixty-one grants for genetics education and newborn screening follow-up services, including grants to seventeen hospital based medical genetics programs, ten pediatric hematology programs, and thirty-four local health departments

Reports from Partners:

Sickle Cell Disease Association (SCDAI) – Talana Hughes informed members that SCDAI has been working closely with the IDPH Newborn Screening Program over the past 5 years, and the agency has a MOU with IDPH to assist in providing follow-up services for children with sickle cell disease or sickle cell carrier status. Each year SCDAI receives referrals for over 300 individuals with sickle cell diseases, including around 100 adult clients, and 3,000 to 4,000 referrals for individuals with sickle cell carrier status. SCDAI has been the recipient of two federal grants related to newborn screening services, and to increasing the accessibility of medical homes for individuals with sickle cell diseases.

Sudden Infant Death Services (SIDS) of Illinois – Annamarie Valdez, the new CEO of the SIDS of Illinois told members that for fiscal year 2011 the organization provided education on sudden infant death syndrome and the importance of providing safe sleep environments for all infants to over 40,500 professionals and members of the general public.

Division of Specialized Care for Children – Tess Rhodes responded to a question about DSCC services by explaining that conditions for which DSCC provides services are determined by the program’s Administrative Rule, and that the criteria for these conditions and services are subject to Rule changes.

Chicago Center for Jewish Genetic Disorders – Claudia Nash reported that DNA Day activities for fiscal year 2011 included promoting awareness of genetics issues, and education about genetics and the importance of family health histories. Genetic counselors gave presentations at high schools, colleges and local health departments. Plans are in progress to continue working with the Center in preparation for the 2012 DNA Day.

The meeting was adjourned at 2:25 p.m.

